APPENDIX B - GLOSSARY

"A" BANDS:

Terminal nucleotide addition occurs when Taq DNA polymerase adds a nucleotide, generally adenine, to the ends of amplified DNA fragments in a template-independent manner. The efficiency with which this occurs varies with different primer sequences. Thus, an artifact band one base pair shorter than expected (i.e., missing the terminal addition) is sometimes seen. Use of more template than recommended can generate incomplete terminal nucleotide addition at some loci.

ADENINE:

A purine base; one of the four molecules containing nitrogen present in the nucleic acids DNA and RNA; designated by letter A.

ALLELE:

One of two or more alternative forms of a gene.

ALLELE FREQUENCY:

The proportion of a particular allele among the chromosomes carried by individuals in a population.

ALLELIC LADDER:

An allelic ladder is an artificial mixture of common alleles present in the human population for a particular STR marker. They are generated with the same primers as tested samples and thus provide a reference DNA size for each allele included in the ladder.

AMINO ACIDS:

The building blocks of proteins. There are 20 common amino acids; they are joined together in a strictly ordered "string" that determines the character of each protein.

AMPLIFICATION:

Increasing the number of copies of a specific segment within a DNA chain. "Building" DNA. PCR DNA analysis uses this technique to amplify specimens that are too small to use for the RFLP method.

ANNEAL:

The process by which the complementary base pairs in the strands of DNA combine.

AUTOSOME**:**

A chromosome not involved in sex determination. The diploid human genome consists of 46 chromosomes, 22 pairs of autosomes, and one pair of sex chromosomes (the X and Y chromosomes).

BASEPAIR:

Two complementary nucleotides joined by hydrogen bonds; basepairing occurs between A and T and between G and C.

BASE SEQUENCE:

The order of nucleotide bases in a DNA molecule.

BLEED THROUGH:

The phenomenon of "bleed through" can occur with incomplete separation of colors. Care must be taken when evaluating light bands to determine if off-ladder bands or minor bands are a result of bleed through. This is easy to determine by checking other layers. This phenomenon occurs when gels are overloaded.

CHROMOSOME:

A discrete unit of the genome carrying many genes, consisting of proteins and a very long molecule of DNA, visible as a morphological entity only during the act of cell division. The entire human genome is tightly packaged into 23 pairs of chromosomes which are located within the nucleus, or center, of the cell.

CLONING:

The procedure for producing identical DNA sequences.

CODIS:

The COmbined DNA Index System. CODIS refers to the entire system of DNA indexes (convicted offender index, close biological relatives index, population file, forensic index, unidentified persons index, and missing persons index) maintained at the National, State, and Local levels.

CODON:

A group of three bases on the DNA molecule that will code for an amino acid, the chemical units of proteins.

CROSS-HYBRIDIZATION:

An interaction of a DNA sequence with another sequence (e.g., probe) that is not perfectly complementary to it. Cross-hybridization occurs at low stringency. See also DNA heteroduplex.

CYTOSINE:

A pyrimidine base; one of the four molecules containing nitrogen present in the nucleic acids DNA and RNA; designated by letter C.

DNA ADVISORY BOARD (DAB):

A board to develop standardized DNA quality assurance methods appointed by the FBI Director in accordance with the DNA Identification Act of 1994.

DEGRADATION:

Partial or complete deterioration of a biological substance by chemical or physical means.

DELETIONS:

Results from the removal of a sequence of DNA, the regions on either side being joined together.

DENATURATION:

Conversion of DNA from the double-stranded to the single-stranded state, usually accomplished by heating to destroy chemical bonds involved in base pairing.

DIFFERENTIAL AMPLIFICATION (ALSO REFERRED TO AS ALLELIC DROP OUT):

The preferential amplification of one allele over another, which can result from incomplete denaturation of the target molecules of one allele. Alternatively, this may occur when one of two alleles is replicated more readily than the other, as in amplification of genetic loci in which some alleles are much longer than others (VNTR region).

DNA HETERODUPLEX:

Double stranded DNA molecule in which the two strands do not have completely complementary base sequences.

DNA PROFILE:

A DNA profile consists of a set of DNA identification characteristics, i.e., the particular chemical form at the various DNA locations (loci), which permit the DNA of one person to be distinguishable from that of another person.

ELECTROPHORESIS:

A technique in which molecules are separated by their velocity in an electric field.

ENZYMES:

Proteins that catalyze specific biochemical reactions, such as *Taq* polymerase, which cause the addition of bases.

EXPONENTIAL AMPLIFICATION:

Replication of DNA in which the copy number of the target sequence approximately doubles in each cycle or round of replication.

FLUORESCENCE:

Emission of or the property of emitting electromagnetic radiation usually as visible light resulting from and occurring only during the absorption of radiation from some other source.

GEL:

Semisolid matrix (usually agarose or acrylamide) used in electrophoresis to separate molecules.

GENE:

A stretch along a chromosome that codes for a functional product (either an RNA molecule or its translation product, a polypeptide).

GENOTYPE:

The total of the genetic information contained in the chromosomes of an organism; the genetic makeup of an organism.

GUANINE:

A purine base; one of the four molecules containing nitrogen present in the nucleic acids DNA and RNA; designated by letter G.

HARDY WEINBERG EQUILIBRIUM:

A principle of population genetics which states that population gene frequencies and population genotype frequencies remain constant from generation to generation if mating is random and if mutation, selection immigration and emigration do not occur. If these assumptions are true, it should be possible to calculate genotype frequencies from observed allele frequencies.

HETEROZYGOTE:

A fertilized egg (zygote) with two different alleles at a designated locus. An individual organism that has different alleles of a particular gene on each member of a pair of chromosomes. An organism is heterozygous to a given gene if its two alleles are different.

HOMOZYGOTE:

A fertilized egg with two identical alleles at a designated locus. An individual organism having identical alleles of a particular gene on each member of a pair of chromosomes. An organism is homozygous to a given gene if its two alleles are the same.

IN VITRO**:**

Outside a living organism

JOE:

6-Carboxy-2',7'-dimethoxy-4',5'-dichlorofluorescein. Fluorescent dye used for STR allele labeling.

LINKAGE:

Describes the tendency of genes to be inherited together as a result of their location on the same chromosome; measured by percent recombination between loci.

LOCUS:

The position on a chromosome at which the gene for a particular trait resides; locus may be occupied by any one of the alleles for the gene.

MENDELIAN INHERITANCE:

The passing of genes from parent to progeny according to a pattern of independent segregation of alleles and the independent assortment of unlinked genes (e.g., genes on different chromosomes) during the formation of gametes.

MICROVARIANT:

Alleles that have a similar intensity to the other major bands for a locus but will not align with the allelic ladder. Alleles with one, two, or three nucleotides shorter than the common four base repeat alleles which are located between two alleles on the ladder are reported in accordance with the recommendations of the DNA Commission on the International Society of Haemogenetics. The number of complete repeat units is represented by an integer and any partial repeat is designated by a decimal followed by the number of bases in the partial repeat. Therefore a band occurring between 5 and 6 alleles and which is 1 bp from the 5 allele is designated as a 5.1

MUTATION:

Any inheritable change in DNA sequence.

NUCLEIC ACID:

A nucleotide polymer that DNA and RNA are major types.

NUCLEOTIDE:

A unit of nucleic acid composed of phosphate, ribose or deoxyribose, and a purine or pyrimidine base.

OLIGONUCLEOTIDE:

Single stranded DNA molecule of two or more nucleotide units in length.

PCR PRODUCT:

The double stranded DNA fragment of defined size and sequence which results from the PCR amplification process.

POLYMERASE CHAIN REACTION (PCR):

An in vitro process that yields millions of copies of desired DNA through repeated cycling of a reaction involving the DNA polymerase enzyme.

POLYMORPHISM:

Difference in DNA sequence among individuals. Genetic variations occuring in more than 1% of a population would be considered useful polymorphisms for linkage analysis.

POPULATION:

A group of individuals residing in a given area at a given time.

PLATEAU EFFECT:

A phenomenon of late stages of PCR amplification in which there is a progressive attenuation in the rate at which target sequence accumulates in each successive cycle.

PRIMERS:

Oligonucleotides which serve as growing points for polymerization of a new strand of DNA along a complementary template strand.

SEX CHROMOSOMES (X AND Y CHROMOSOMES):

Chromosomes that are different in the two sexes and involved in sex determination.

SHORT TANDEM REPEATS (STR):

Multiple copies of an identical DNA sequence arranged in direct succession in a particular region of a chromosome.

STOCHASTIC FLUCTUATION:

A phenomenon occurring during the amplification of low levels of DNA resulting in an unequal sampling of the two alleles present from a heterozygous individual.

STUTTER BANDS (SHADOW BANDS):

Some STR loci have a tendency to produce one or more minor PCR bands which are typically smaller than the major allele by 1 (n-1), 4 (n-4), or 10 (n-10) bases, depending on the locus. Generally peak height can readily distinguish true alleles from "stutter" peaks.

THYMINE:

A pyrimidine base; one of the four molecules containing nitrogen present in the nucleic acids DNA and RNA; designated by letter T.

♦**END**